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## **Mitochondrial donation, patient engagement and narratives of hope**

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### **Abstract**

This article develops the sociology of hope and patient engagement by exploring how patients' perceptions and actions are shaped by narratives of hope surrounding the clinical introduction of novel reproductive techniques. In 2015, after extensive public debates, the UK became the first country to legalise mitochondrial donation techniques aimed at preventing the transmission of inherited disorders. The article draws on the accounts of twenty-two women of reproductive age who are at risk of having a child with mitochondrial disease and would be the potential target of the techniques. We explore the extent to which our participants engaged with the public debates and how they accounted for their support of mitochondrial donation. We show that while the majority of our participants were in favour of legalisation, they did not necessarily wish to use the techniques themselves. We found that hope was multi-faceted, involving hope for self, hope for family and hope for society. We conclude by considering the implications of hope narratives for patients and families and the important but potentially limited role that patients can play as advocates for technology.

## Introduction

Set in the context of public debates on ‘mitochondrial donation’<sup>1</sup>, this article develops the sociology of hope and patient engagement by exploring how patients’ perceptions, actions and identities are shaped by narratives of hope. Techniques of mitochondrial donation can prevent the transmission of maternally inherited mitochondrial disorders by conceiving an offspring with the genetic material of three people. These high-profile techniques were legalised in the UK in 2015 following extensive parliamentary debates, public consultations and meetings, institutional reviews and calls for evidence. There have been great expectations surrounding these techniques, which have been reflected and magnified through these debates. This article identifies these narratives of hope as ubiquitous, where the technology is framed in terms of potential treatment for people affected by mitochondrial disorders, and as associated with a powerful discourse of the prevention of suffering.

We draw on interviews with women who are the imagined target of the techniques, women of reproductive age who are at risk of transmitting maternally inherited mitochondrial disease, and analyse how the hope narratives surrounding mitochondrial donation have been perceived and constructed by these women. Indeed, while much existing work has analysed the strategic uses of hope narratives at an institutional level, less attention has been given to what Petersen and Wilkinson call the ‘performativity of hope’ (2015: 117), that is, the ways these discourses are interpreted by patients and how they influence their views, identities and actions in practice. The question we address is why participants supported the legalisation of techniques, and indeed, invested their time and energy in the campaigns to legalise the techniques, when they might not be planning to use the technique themselves? Overall, we ask what does hope mean for patients in the context of emerging reproductive technologies and what are the implications of such powerful discourse?

In this article, we first look at patients’ perceptions of mitochondrial donation and highlight the role of hope in patients’ support for their legalisation. Hope is multi-faceted and the reasons why participants supported the techniques are more complex than simply that they want to use the technologies themselves. We then describe how patients’ engagement in the debates has been shaped by this narrative of hope. We end with a discussion of the implications of hope narratives for families affected by mitochondrial disorders. Whereas previous conceptualisations of hope in relation to patient activism have highlighted resistance to the establishment, the way our participants have contributed to these narratives is by acting

as ‘advocates’ for the technology, a role which has more limited potential to influence the research or policy agenda.

### **Legalising mitochondrial donation and the central feature of hope**

Mitochondria are small structures contained in our cells which provide energy. Mitochondrial disease can be caused by faults in the mitochondria, and can produce a wide range of symptoms including diabetes, epilepsy, muscle weakness, severe fatigue, heart problems and difficulties with balance and walking. Faults can be caused either by mutations of nuclear genes or mutations of mitochondrial genes, each leading to different patterns of inheritance. In this article we focus only on maternally inherited mitochondrial disease, the prevalence of which is difficult to estimate due to the wide range of clinical features, variable correlation between symptoms and genotype, and differences in how and when individuals seek medical care. The current estimated prevalence rate for maternally inherited mitochondrial disease is 1 in 5,000 (Gorman et al. 2015). As mitochondrial genes are inherited through the female line, both sexes can be affected by maternally inherited mitochondrial disease but it is only females who are at risk of passing on those faults to her children. Many people can live without realising they have faults in their mitochondria, while for others it can be serious and sometimes fatal. There is no cure and treatment is limited. Scientists at the Wellcome Trust Centre for Mitochondrial Research in Newcastle, UK, have developed a specific IVF technique to avoid the inheritance of *maternally inherited* mitochondrial diseases involving cell reconstruction using part of a donated egg. The major implication of this technique is that the resulting baby would be born with nuclear genes from the intending mother and father, but their (healthy) mitochondria would come from the donor.

The technique is controversial mainly because the mitochondria genes from the egg donor would be inherited by future generations. This is therefore considered a germ line technology, which had been banned in the UK, and the law needed to be changed before the techniques could be used in practice. From 2012, there was an extensive process of consultations, debates in the houses of parliament, and safety reviews to explore the implications of legalising mitochondrial donation (Dimond 2014). The consultations invited contributions from publics and patients, and concluded that the techniques were broadly ethical (Nuffield Council on Bioethics 2012), that there was general support for the technique (Department of Health 2014) and that the science was ‘not unsafe’ (HFEA 2014). The law was brought into

force towards the end of 2015 (HFEA 2015b) and by the end of 2016, clinics were able to apply for a licence if they could demonstrate expert use of the technology. At the time of writing, Newcastle is so far the only UK clinic licenced to offer the procedure to patients.

As a novel technology, the debates about mitochondrial donation related to several broad concerns including safety and ethical implications. But while these issues were acknowledged and explored at length, the potential benefit to patients was rarely questioned (Herbrand 2017). Instead, the mitochondrial debates could be characterised by a dominant and enduring narrative of hope about the perceived needs of patients. A key part of this narrative was the presentation of mitochondrial disease as being serious or fatal, and of the suffering of patients. Both the debates in the House of Commons and House of Lords for example opened with the same statement about the potential benefits to patients:

The techniques provided for by these regulations offer the only hope for some women who carry the disease to have healthy, genetically related children who will not suffer from the devastating and often fatal consequences of serious mitochondrial disease. (Jane Ellison, House of Commons debate 3 February 2015, Column 160)

Throughout the debates, mitochondrial donation was framed in two main ways. First of all, mitochondrial donation was positioned as a viable, essential and desired technological solution and the ‘only hope’ for some women. Within this framing, there was little room for more clearly defining the category of who might be ‘at risk’, and providing a more nuanced account of the role of reproductive technology in their reproductive decision-making processes. Secondly, mitochondrial donation was positioned as a technology that would prevent suffering, and the suffering of children in particular. As Buchbinder and Timmermans (2014) have highlighted, the mobilisation of suffering can become an important part of political projects because of the emotional response it gains from policy makers. Accounts which feature the ‘desperateness’ of those who could potentially benefit from scientific progress have persuasive appeal (Franklin 1997). Many of the consultation reports and debates highlighted the patient perspective, often focusing on emotional stories about children who have short but difficult lives affected by mitochondrial disease. This emphasis accorded with the activities of the patient group who rose to prominence during the time of the debates. This patient group mostly represented families whose children were affected by mitochondrial disease and presented a particular kind of ‘patient’ experience – healthy women with seriously ill children. People affected by mitochondrial disorders were represented as a homogenous group, sharing a common experience of illness, suffering and

loss, and this informed dominant representations in media coverage, reports and subsequent debates. The power of these particular narratives in the public sphere has led to a narrow representation of patient experiences and has informed a limited vision of what mitochondrial disease means to those affected.

## **Narratives of hope**

In recent years, the concept of ‘hope’ has often been employed in studies of health, illness and medicine. Hope is framed as an ‘emotional attitude’ (Simpson 2004: 428) which ‘orients the hoper towards particular phenomena (in the future)’ (Brown et al. 2015: 209). However, as Petersen and Wilkinson (2015) point out, hope is still too often addressed and understood at an individual level and from a medical or psychological perspective as a valued attitude to promote adherence to treatment regimes or to adapt to changes in health and illness management. Instead, Petersen and Wilkinson (2015: 114) encourage the development of the sociology of hope by ‘critically questioning the values incorporated within the social representation of hope as a positive attribute of health or as a goal of medicine’.

In this respect, a small but growing body of sociological work has highlighted the institutional and cultural processes generating and disseminating hope, as well as their political or economic consequences (Brown 2005, Good 2001, Novas 2006). In particular, social scientists have examined how ‘rhetorics of hope’ are deployed and mobilised, often strategically, to legitimize and promote new research, technologies or treatments, as well as to attract support or funding (Martin et al. 2008, Mulkay 1993, Petersen and Seear 2011). These rhetorics of hope reflect an ‘implicit model of scientific progress’ (Mulkay 1993: 725). Not only does it present scientific innovations and more broadly the relationship between science and society in a positive light, but it implies a forward looking attitude towards future potential benefits which have yet to be realised (Brown 2003, Moreira and Palladino 2005). Social scientists have also looked at how hope works in practice, especially how patient experiences can be affected by hope in their everyday life (Brown et al. 2015, Elliott and Olver 2007). Hope is particularly prominent in the field of reproduction when the desire to have a child, and of the new life which may come with it, generates and justifies high expectations and investments. For individuals facing difficulties to conceive or the risk of transmitting genetic disorders, the technologisation of reproduction conveys the promise of desired and healthy babies, and more broadly the promise of restoring a ‘natural order’ and

life expectations that have been disrupted (Throsby 2004). Reproductive technologies, such as IVF (in vitro fertilisation), PGD (pre-implantation genetic diagnosis), gamete donation or egg freezing thus become ‘hope technologies’ (Franklin 1997: 176, Franklin and Roberts 2006): they keep hope alive against the odds and give patients the possibility to act on their situation. However, the hope generated by scientific and technological progress may confront intending parents with frustrating and disappointing ‘corpo-realities’ (Brown 2005), when bodies fail to comply with technological means. But as Szweczuk (2012) points out, it is precisely the uncertainty regarding the successful outcome of these technologies that enables hope to appear and remain. Franklin describes how women and couples trying to have a child using IVF therefore need to carefully ‘manage’ their hope, whilst caught in cycles of optimism and disappointment. It requires ‘balancing sufficient measure of hope against a realistic appraisal of the likelihood of failure’ (1997: 158), in order to be able to continue and cope with this emotionally laden procedure (Haimes 2013). Ultimately, attempting conception with assisted reproductive technologies appears compelling for many individuals as it enables them to ‘exhaust’ this hope and prevent future possible regret (Tymstra 1989).

Investment in hope can also play a crucial role in generating actions at a more collective level. Novas argues that the hope of cure or treatment plays a crucial role in the formation of patient activism. The ‘political economy of hope’ (Novas 2006: 289) contributes to a transformation of biomedical research and the bioeconomy, in particular through the regulation of research trials and commercialisation of donated tissues. While the notion of ‘political economy of hope’ was initially used to conceptualise the hope produced within the clinical encounter by the clinician, particularly when disclosing a cancer diagnosis (Good et al., 1990), Novas emphasised the economic and political investment in hope and the mobilisation of patient activities. Within this model of hope, patienthood is conferred with agency. Patients are recognised as experts and activists in medical and scientific spaces beyond the clinical encounter, and play key stakeholder roles.

However, as many authors have shown, there are difficulties associated with mobilising hope. There can be a lag between the hope associated with the development of treatments and cure, and their realisation as clinically applicable and available to patients (Brown 2003, Stockdale 1999). There is also the ‘danger’ of risks associated with new technologies. Petersen and Wilkinson highlight how the ‘the power of hope’ has the potential to mobilize diverse constituencies and to engender commitment to clinically unproven treatments or care regimes that may, in time, be shown to have no benefit and may even be dangerous’ (2015: 116). As

they also point out, understanding hope only in terms of whether hope is ‘realistic’ or ‘false’ prevents us moving forward to an understanding of the ‘politics of hope’. In accord with Petersen and Wilkinson, we have directed our attention to the politics underlying mitochondrial donation and its debates as particular ‘hope-promoting’ practices. Indeed, examining the ‘hype’ around mitochondrial donation is important because the debate engaged so many people and appeared applicable to so many, despite mitochondrial disease being a rare disease, and those able to take up reproductive options being an even smaller sub group. Whereas health professionals can play an important role in shaping patients’ responses towards hope (Simpson 2004), our emphasis is the public and political nature of the mitochondrial debates which enabled patients to be positioned as contributors at one particular moment in the developmental trajectory of a reproductive technique.

## **Methods**

This article draws on data collected for two separate research projects focusing on the experiences of women affected by mitochondrial disease. The projects’ aims were twofold: firstly, to understand individual and familial experiences of diagnosis and disease and secondly, to explore their perspectives regarding reproduction options, in particular about the IVF techniques involving mitochondrial donation. For both projects, in-depth interviews were conducted mainly with women of reproductive age (classified in both studies as 18 – 45 years) who were at risk of transmitting mitochondrial disorders to their children.

Project 1 involved interviews with 42 people. Twenty-eight of them were women affected by mitochondrial disorders, either because they were carrying mitochondrial mutations and might develop the disease ( $n \sim 11$ ), or because their child was affected by the disease ( $n \sim 17$ )<sup>2</sup>. Fourteen family members were also interviewed, including partners, mothers and sisters. Patients were recruited through the national patient cohort database held at Newcastle University and a national support group, and ethics approval was granted from De Montfort University and the London NRES Committee. Project 2 involved interviews with 31 people. Eleven of whom were women of reproductive age who had been diagnosed with maternally inherited mitochondrial disease, 11 women were older (over 45 years old) and were diagnosed with maternally inherited mitochondrial disease, and 9 were partners or male relatives. Patients were recruited through the national patient cohort database held at



Newcastle University and ethics approval was granted from the North Scotland NHS Research Ethics Committee. Only the interviews conducted with 22 women of reproductive age who carry *maternally* inherited mitochondrial mutations are reported here, i.e. 11 from each project who were under 45 and at risk of transmitting mitochondrial mutations). The ages of the women ranged between 19 and 44 and they all lived in the UK.. The women came from diverse social and economic backgrounds, presented various professional and family situations and ten participants had children at the time of the interview, including three who had their children after being diagnosed with mitochondrial disorders.

Although the women presented different symptoms and health conditions, ranging from no symptoms to moderate symptoms threatening life quality, they shared the risk of having a child with mitochondrial disease. These women are potential future users of mitochondrial donation and have been presented through the debates as ‘patients’ in a broad sense. They also either identify as ‘patients’ with mitochondrial disease, or consider themselves, or their children (or future children) as being at risk of developing mitochondrial disease. They therefore have, in principle, the same reproductive options if they want to have a child: have a child without assistance with a risk of transmitting the disorder, adoption, egg donation, prenatal diagnosis, pre-implantation genetic diagnosis, and possibly, mitochondrial donation (Richardson et al. 2015). In this article, we only discuss participants’ views on mitochondrial donation and not their responses to other available reproductive options.

Most of the interviews took place in the respondent’s own home, and lasted between 45 minutes and 2 hours. The interviews were audio recorded and transcribed. Data were analysed using thematic analysis (Braun and Clarke 2006), which involved reading the transcripts and identifying key themes. Once analysis was conducted on each data set by the corresponding author, the authors met to talk about the themes which were prominent in each data set and where comparisons could be made. We shared and re-analysed together the relevant parts of the anonymised transcripts. The key themes which are discussed in this article, including expressions of support for mitochondrial donation and contributions to the debates were prominent across both data sets.

With our combined findings, this constitutes the largest sample of women carrying mitochondrial mutations to be studied from a qualitative perspective, as well as the first sociological analysis focusing on the experiences of these women following legalisation of mitochondrial donation in the UK.

## **Supporting mitochondrial donation**

Amongst our participants, only three women did not overtly support the legalisation of the techniques. Two of the women took this position because they suggested that they did not know about the techniques and the other for religious concerns. The rest of our participants explained at length why they supported the legalisation of the techniques. However, they did not necessarily support mitochondrial donation because they wanted to use the techniques themselves. Some participants were not planning to have children, or any more children, sometimes because they felt their own health threatened their ability to look after a family. Nonetheless, whether or not women have already had families, were planning to start a family or have decided against having children, most participants expressed that they supported the techniques. Their stories highlight the various facets of hope which were represented by the techniques. We have grouped and developed these under the following themes: hope for self, hope for the family and hope for society.

### *Hope for self: the potential to have healthy children*

For a minority of our participants, the techniques represented a very real opportunity to improve their own chances to have unaffected children. The techniques and their legalisation were perceived as part of their own journey. Beth [C22]<sup>3</sup>, who was in her early twenties and in a relationship for 3 years, explained why she would want to use the techniques:

Well I don't want to have a child that has any [faulty] mitochondria at all, just because I think it would be hard for me showing symptoms and things like that. I think it would be hard for me to then deal with a child that has the same as what I have, you know, with my energy levels and things like that it would be really hard for me to cope. I wouldn't want to have to go through what we went through with Tom [her brother who died from MT disorders] again and I just think it would be... I just would like a healthy baby (laughs).

This woman's account is embedded in her experience of living with the increasing signs of the disease. Although she works full time, she was beginning to experience extreme tiredness and much lower 'energy levels'. She was interested in mitochondrial donation as it was very important to her to have a genetically related child later on. However, she expressed that she

definitely did not want to pass on the disorder and allow her child to go through what she herself experienced. She also did not think she would be able physically to cope with a child who was disabled, as she often felt weak and could see how much energy it required when her mother had to take care of her brother who was affected by mitochondrial disorder and died a few years earlier. Mitochondrial donation was therefore a means to avoid reproducing the still deep and traumatising experience it had been for her family.

Another woman who saw the techniques as allowing her to have a healthy child was Alice [C13], late twenties, who was interviewed with her boyfriend. After her mother was diagnosed with mitochondrial disorder, she herself was tested for the mutation and learnt that she carried the faulty gene, though she had not experienced any symptoms at the time of the interview:

In a way, with everything being so doom and gloom about the diagnosis, it seemed like a bit of a ray of light at the end of the tunnel. Because it would give us the opportunity to live a normalish life, even though we are a family affected by mitochondrial disease, we will still be able to have a family and be normal. Knowing that our babies [are] not going to be affected by it.

For Alice, mitochondrial donation represented an opportunity for her to have a healthy child and therefore ‘live a normalish life’ in the context of a severe illness which had affected other members of her family. Whereas Beth’s account was primarily contextualised within the experience of degenerative disease and Alice was not currently displaying any symptoms of disease, they both supported the legalisation of mitochondrial disease because it represented an opportunity for them to have healthy biologically-related children. Thus mitochondrial donation was viewed as a viable technology, whether or not the woman was experiencing symptoms of mitochondrial disease.

While some respondents who expressed support for the techniques did so *because they believed they were in a position to use them*, this group of women were in a minority. They were the younger participants of our sample who had not yet children. For the majority of our respondents, there was not such a direct link between supporting the techniques and wishing to, or being in a position to use the techniques themselves.

#### *Hope for the family: the potential for daughters to have healthy children*

Respondents spoke about the future benefits of mitochondrial donation when their daughters would be preparing to have children. This was the case for Helen [R16], who was in her early

forties and had diabetes and deafness. She was diagnosed with mitochondrial disease when she went for genetic testing with her mother and sister, after they developed diabetes. By that stage, she had had a son and daughter. Her daughter was 10 years old and appeared healthy at the time of the interview. Speaking about mitochondrial donation, she explained:

It's not going to affect me in any way. It's more important for me because it's going to affect me directly in terms of my daughter, it's going to affect her very directly. At the end of the day the reproductive thing offers me nothing, it offers my daughter hope, it doesn't offer me anything.

Another participant, Julie [R10], also said she supported the techniques because it offered hope for her two daughters who were both born after her diagnosis. Julie was in her early forties, and was diagnosed about five years ago through her hearing problems. She said that her mother had not yet been diagnosed, although was thinking about it because they had a large extended family and that information might be relevant to others in the family. At the time of the interview her two daughters were in their early teenage years:

So I've definitely taken more interest in [the debates about mitochondrial donation] because I know that could affect both of them two as they go along, sort of thing. And hopefully, by the time they are at the age for having children, it'll be all sorted and it's an option for them. I mean, then that will be their decision, won't it, but it's there for them at least to have, isn't it?'

Another woman, Sally [C23] explained that if she could, she would like to use mitochondrial donation later on to have children. She was in her early twenties, did not have any children yet and although she felt healthy, two of her siblings were suffering quite seriously from mitochondrial disease. Her concern was not so much to avoid the transmission of the disorder but rather to guarantee reproductive health to her children. To her, the techniques represented an opportunity to protect her future children from carrying the psychological burden and the medical risk of transmitting the disorder to future generations:

I don't want my child to have to think how they're going to have a baby because they might pass it on to their children, like it's not a nice thing to think about. So I don't want to conceive naturally due to that reason only. Like my levels at all don't bother me, because I know that I'm not affected, but I just don't want it to carry on for generations, so I'd much rather it just stop here and be done with.

Question: So you would be willing to use the technique as well?

Yes, just because I wouldn't want to pass it on and for a child to grow up with those worries about am I going to get worse? Is it going to be ok? Am I going to pass it on to my child? I just think it would be easier.

Sally provides an interesting account because, like many other respondents, she focuses on several aspects of risks. She not only discusses her reproductive choice in terms of the child inheriting the condition but importantly, she also describes her concern about passing on reproductive risk to her own child. What unites the woman and her future (female) child is that both would experience the possibility of making potentially difficult reproductive choices. It also suggests that parents are not just concerned about the child's genetic risk more than their own health (McAllister *et al.* 2007), but also about the child's future reproductive risk.

The patients in our sample presented various reasons as to why their children (or future children) would benefit from mitochondrial donation, including offering an additional reproductive option and allowing daughters to have children without fear of having the disease. What is interesting about this position is that several of our participants themselves were not diagnosed until after they had had children, and therefore are projecting a particular future for their daughters, where risk is known and is a key factor in reproductive decision making. This suggests how our participants' projections for hope in the future are based on their reflections about their own experience of having children but combined with the new knowledge of what it means to live with illness or reproductive risk.

#### *Hope for society: supporting opportunity for others*

The previous sections have highlighted how some participants expressed their support for mitochondrial donation because of the direct benefits to themselves or their families. But overall, the majority of our respondents remained committed to the idea that mitochondrial donation should be made legal and should be made available. In doing so, they highlighted the wider benefits of such technologies in terms of supporting the reproductive choices of other women, and for society.

It was clear that participants recognised the need to support the wishes of others to have the opportunity to have a healthy child, for example, those who might have experienced the heartbreak of having a child die because of mitochondrial disease. For instance, Wendy [C1] did not know whether mitochondrial donation would be legalised and available in time for

her to use it in order to have a family as she was already in her late thirties. In any case, she thought it should be authorised as it would help other people and give them hope:

From what I have read, I think it would be a good thing for future people to have their own child. They probably feel like me because of the difficulty of carrying the mitochondrial disease, it does give people hope out there. [...] It prevents that fault being passed down to another child in the future. To me, it could decrease the amount of people that are actually ending up with the mitochondrial disease in the future.

This was also the case for Ruth [R22], who is in her late 30's and was diagnosed about 7 years previously. She is mainly affected by migraines and tiredness, and is the only member of her family to have been diagnosed, and the only member to show symptoms. Although she does not have children herself, she accounted for her support for the techniques by highlighting the suffering of parents who risk losing their children to the disease:

I just think if you can [stop] parents going through losing children or children having this disease, then I'm all for it.

Overall, these accounts illustrate the ubiquity of hope in patients' perceptions of mitochondrial donation. Participants supported the legalisation of mitochondrial donation for a number of reasons, including for personal reasons. But supporting the technologies on behalf of others presents a different account in terms of the narrative of hope. It provides a reminder as to why we need to pay attention to the way in which technologies are represented in the public arena. Presenting mitochondrial donation as a technology which can stop the suffering of children (in particular) remains a dominant trope, even for those who were without symptoms themselves and those who experienced mitochondrial disease as adult onset.

### **How is patient agency mobilised by hope?**

Not only were most women we interviewed in favour of mitochondrial donation but most of them also actively supported them by becoming involved in the related debates. Engagement took very different forms: following the debates on television or radio and reading newspapers; asking friends to text the result of the votes; writing to their MP and sometimes meeting with him or her; attending public meetings as a participants or speakers; attending Parliament during the voting process; speaking to the media about their experience of living

with mitochondrial disease and about their views on mitochondrial donation; responding to calls for evidence; taking part in social events to collect funds, offering to take part in research, including for example, and at the extreme end, offering their eggs for donation for research purposes.

One of the key ways that patients could contribute to the campaign, which patients were actively encouraged to do, was to make contact with their MP before the vote in the House of Commons, either by meeting and talking to MPs during special sessions organised by the support group before the key debates at the Parliament, or by sending pre-printed support letters prepared by the clinic to their MP. One participant, Anna [R14], was interviewed with her partner and spoke at length about how she had contributed to the debates, mainly by contacting her MP to ask for their support. Anna was diagnosed after her mother was diagnosed – her mother had become ill quite suddenly, and her diagnosis therefore came as a shock to her and her siblings. Anna did not have children but was currently investigating different possibilities through assisted reproduction:

[The director of a specialist mitochondrial disease clinic] sent us a letter that we could forward on. And actually, it was a well written letter and it wasn't saying 'This is the best thing ever'. It was 'Please support the debate'.

The accounts of our respondents who actively contributed to the debates highlight that the work of the engaged patient does not just entail making contact with their MP. Many respondents followed up whether or not the MP voted in favour. One participant, Jackie [R8], who was in her late thirties, and was diagnosed five years previously after suffering extreme tiredness, describes the extent to which she went to make contact with her MP:

I spoke to my MP about it. She voted for it. I spoke to her the night before the vote, because I left messages and we were due to meet but the meeting was after the vote. So I called them Monday morning and I said, 'Look, there's no point in us meeting later in the week if the vote is tomorrow, can she call me today?' And she didn't know anything about me. So she called me on Monday at nine o'clock in the evening, before the vote. And she [her MP] said she was inclining to vote for it but after talking to me she was determined to vote for it.

Two of our participants who were asymptomatic also actively engaged by attending the public meetings which took place at Parliament, and telling the audience how important it was for them to access mitochondrial donation in order to have healthy children. Moreover,

several participants spoke to the media to tell their stories and to manifest their support to the techniques.

What unites all these accounts and the diverse activities of patients is that patients were very much emotionally engaged. One of our participants [C9], after following the vote in the House of Lords, told us that she ‘started crying that day, I was so happy. It’s a good thing’. Aligned with the opportunity to provide the patient voice to the debates, is the fact that overall their contributions were highly valued. Patients were able to be presented as having ‘direct experience’ of the disorders and being potential ‘users’ of the techniques, and as such, they provided evidence of the desirability and essentiality of mitochondrial donation. Engaging in the debates has been empowering for some participants, as was the case for Alice [C13] mentioned earlier:

He [the director of a specialist mitochondrial disease clinic] said [the techniques] will go through Parliament shortly but he didn’t sort of try and put our hopes up and say ‘well this technique will be available for you’. He just said this is something like ‘we have been working on and it is going to go through Parliament shortly’. And that was how it was left until he contacted us and we thought ‘oh god it’s come round’. And we were glad in a way to be part of it thinking well this is potentially something that is going to affect our future. And if we can help in any way then we will so we were honoured, well I was, that he asked us to help because it’s sort of being part of history in a way, isn’t it.

Being part of the debates had important implications for Alice, particularly because it allowed her to be ‘part of history’. For Katie [C9], a young single woman, the debates presented a public stage as an opportunity to ‘come out’ and embrace her diagnosis and her experience of illness. She has started to develop several mild to severe symptoms over the past few years. She wants to have children later on, though she knows her condition is worsening and she is worried about the extent to which it will get. She does not want to pass on the disease, so she thought first about adoption, but then she became interested in mitochondrial donation. After hearing about the debates, she told her story to the media, which was the first time she openly talked about her experience, even to friends:

I had some school friends messaging me on Facebook saying that it was really admirable of me to come with my story. When I was in school they didn’t even know, the teachers didn’t know, because I would be very slow in Physical Education, I



would get very tired I wasn't really like everybody else. But now this has all come out, it's probably because I am mitochondrial that's why I was getting really tired. But it's all come out now and I think they do understand a bit more about my condition and hearing. Not many of my friends know much about it. [...]

Question: And you were not afraid that your friends and everyone would know then that you had a disorder?

I wasn't afraid I thought everyone should know now. [...]

I don't mind telling people about it, so they have more understanding about how I feel and how it's affecting me.

Whatever the nature and extent of their involvement, it is important to emphasise that the mitochondrial donation debates presented the first opportunity for most participants to become involved in activities on a public stage and patients took up the opportunity with enthusiasm and passion. Thus the mitochondrial donation debates remind us of the importance of the political context when we are examining hope (Novas 2006). The mitochondrial debates follow a format which is increasingly familiar when discussing novel technologies, where the technology is framed as what patients want, and the patient experience is showcased as support. Indeed, we have found that patients have widely supported the techniques, and taken up the opportunity to express their support. But it is important to note, in the context of examining patient activities around the central theme of hope, that these opportunities to speak in favour of the technology and its legalisation arose from the nature of the debates (for example, the numerous calls for evidence and media interest), rather than being identified as opportunities by the patients themselves. Mobilised by the clinics and the support groups, patients in the mitochondrial debates were framed as users and as drivers of the technology, but they were invited to take up this position rather than design the role for themselves.

## **Discussion**

The debates about mitochondrial donation were dominated by the culturally powerful narrative of hope, bolstered by stories about patients and by the mobilisation of patients themselves. Patient experiences became important evidence in support of the legalisation of the techniques, and patients enthusiastically took up the opportunities to contribute to the debates. Patients (or parents of an ill child) were given a high status as experts within the debates. They were allocated the opportunity for their voices to be heard, sometimes over

others, because of their status as potential users. Whether speaking for themselves or spoken for on behalf of others, patients ultimately played a key role in the mitochondrial debates because their stories were persuasive about the difficulties of living with mitochondrial disease, and by association, of the value of mitochondrial donation and its legalisation.

But we have found that the mitochondrial debates were limited in scope, particularly around how the patient experience was presented. Mitochondrial disease is a complex condition, highly variable, and for many people will be late onset. The representation of mitochondrial disease as only affecting seriously ill children does not do justice to the experiences of many of those who are currently living with mitochondrial disease as adults. In addition, mitochondrial donation was widely represented as a technological solution, with little discussion in the debates about how women with mitochondrial disease make reproductive decisions and might negotiate reproductive options. But importantly, we found that our participants did not express concerns about these discrepancies. Instead, the majority of them expressed support for mitochondrial donation and their accounts suggested the extent to which they were willing to emotionally engage in the campaign to legalise the technology.

Our examples show that participants supported the techniques for a number of reasons. For them, hope is complex and multifaceted, involving hope for self, hope for family and hope for society. Some of our participants invested in the technology because they believed that the techniques could be personally beneficial. Mitochondrial donation appeared as an important route for them to have a child in the near future. Their accounts reflected an immediacy associated with the experience of mitochondrial disease, and their stories in the most part reflected those in the media accounts of women who have either suffered the loss of a child or have a severely ill child. In this context, participants felt that the techniques would provide them with the opportunity to have a healthy child. In contrast to their hope for self, another kind of hope emerged as prominent through our research. This is where women expressed the hope and expectation that their own daughters would be able to use the technology, and allow them the option of having a healthy child without fear of passing on the disease. Hope for family represents a move away from personal experiences of health and reproductive options, and instead focuses on the (future) reproductive options of children or future children. In comparison to the dominant narrative that women will use mitochondrial donation to have their own healthy genetically related baby, the nuanced reasons as to why women might support the legalisation of a reproductive technology are much less represented within media accounts. It also represents an important field for future research about how and

when families communicate about reproductive risk to their children. Finally, many of our participants expressed their support for the techniques because of the benefit they could bring to other families, who might be more seriously affected and might have had severely ill children. In this case, even when the stories which appeared in the media or public accounts did not represent their own experiences, our participants supported the techniques by empathy for other people.

Despite their various acts of engagement, it is important to highlight that none of our participants could be considered as ‘activists’ fighting *against* the institution in way that Taussig and colleagues (2003) amongst others have described. ‘Patients’, understood in a broad sense, were allocated a space to become involved and detail their positions, rather than having to fight for the right to speak. In this respect our work contrasts with what is known or assumed about patient activities in the context of the political economy. Novas highlights how ‘patient organisations not only challenge the authority of biomedical research but also help to transform the contexts in which it takes place’ (2006: 291). The mitochondrial debates provide an example of how the political activities of patients do not necessarily challenge authority. Instead, the patient role became more like that of an ‘advocate’, or as a witness, explaining the daily implications of their experiences of being affected by mitochondrial disease, and providing personal testimony as to how and why legalisation is essential. This suggests a more limited role for patients in influencing the research agenda.

According to Novas (2006: 292):

Science constitutes one horizon along which potential futures are constructed. By engaging with scientists and advocating particular forms of research, treatment modalities and forms of regulation, patients’ organizations are actively involved in shaping particular futures to the exclusion of others.

The potential for patient organisations’ activity to exclude other futures is an important aspect to apply to the mitochondria example. The involvement of patients has helped to shape the biomedical future in the case of mitochondrial disease, but it might not be a future that patients would choose if they were allowed greater political power. There is no cure for mitochondrial disease and treatment is limited because of its genetic complexity (Rai et al. 2015). Even so, at the time of the mitochondrial donation debates, the possibilities for developing treatments had not emerged as a political issue, and we have not yet witnessed large scale activities, requiring such extensive patient involvement, around the development

of a cure for mitochondrial disease for those living with disease rather than preventing the ill-health of future others.

The case of mitochondrial donation suggests that while patients have wide opportunities for engagement, they have a narrow range of political options. Their options were shaped by the technologies currently being developed (reproduction rather than treatment) and the political will of the scientists and clinicians who are developing them. Patient power, in the case of mitochondrial donation, was restricted to public engagement to support the legalisation of one technology which has limited potential to change patients' current lives.

Following the debates, it seems that the trajectories of the clinical team, and the hopes of patients and families, were only momentarily aligned, and for a particular purpose. While the debates gave patients the opportunity to have their voice heard and to become publically engaged by supporting the legalisation of the techniques, for most of our participants this involvement was short term. One reason for this is that campaign activities stopped quite abruptly after the legalisation of mitochondrial donation. Indeed, the adoption of the law in February 2015 was followed by a long licencing process which was only completed in October 2016. During this period, the support group was not required to be so politically visible while they waited for the procedures to be put in place.

Alongside the legalisation of mitochondrial donation, the mitochondrial debates have no doubt increased public awareness and raised the profile of a rare genetic disease. But it is important to remember the social significance of particular representations of disease and technological solutions. The dominant representation of a disease that causes child suffering and early death did not appear to distress our participants, but it might have implications for others living as adults with the disease and their family members. In addition, while mitochondrial donation was represented as a technology that all patients could use, it is only suitable for patients with maternally inherited mitochondrial disease which would rule out a large proportion of the patient population with different kinds of mitochondrial disease (such as those with recessive or dominant inheritance patterns). And of course, it would not be accessible to women who do not know that they are at risk of having a child with the disease. However, with the recent licencing of the techniques, it was also not clear whether some in our sample would be able to access the techniques, even though they may think so. The new regulation will indeed include a clinical judgement about their risk of having a child with the condition and the potential severity of the future child's symptoms. In short, it must be assumed under current legislation and licencing procedures that the diagnosis of maternally

inherited mitochondrial disease does not necessarily mean that someone will legally be able to use the techniques. This suggests that there is potential for some to experience a ‘false sense of hope’ (Stockdale 1999: 87). This article contributes to the sociological understanding of hope by highlighting how patient voices were welcomed and valued within a highly public and politicised debate. But by identifying how patients played the role of advocates of the technology, we also highlight the potential vulnerability of patients. Our study provides empirical evidence of the tensions inherent in public debates and we encourage researchers to continue to question how patient voices are registered and enrolled as imagined users or advocates of future technologies.

If the combination of hopeful technologies with the power and currency of patient support is to become the norm for public engagement strategies around the introduction of novel biotechnologies (as is starting to be the case with genome editing), then our article provides evidence that we need to think more carefully about patient contributions. We also ask therefore for social scientists to think about how future visions are constructed and how patient activities are directed towards specific agendas. Who is directing, and who or what is excluded, particularly when technologies are not going to improve the health of those living with disease, from the political economy of hope is worthy of further research.

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## **Notes**

1. We use the term mitochondrial donation throughout this article as this was the main term used in UK institutional reviews (Department of Health, 2014; HFEA, 2015a), but we acknowledge that there is divergent views over the accuracy of the term (see for example, Haimes and Taylor 2017, Dimond and Stephens 2017).
2. Most of these children’s disorders were caused by nuclear defects. This means that their mothers are not suitable to use mitochondrial donation.

3. We have used pseudonyms to refer to our participants, in addition to their reference number. 'C' refers to project 1 and 'R' to project 2.

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